Fetal Alcohol Spectrum Disorder—Issues of Misdiagnosis and Missed Diagnosis in Black Youth: A Case Report

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ABSTRACT

Fetal alcohol spectrum disorder (FASD) is an umbrella term used to describe the set of conditions that result from prenatal alcohol exposure (PAE) that lead to cognitive impairment, neurodevelopmental delays, socioemotional and behavioral problems, medical complications, and/or secondary disabilities. In addition, various internalizing and externalizing disorders share similar symptoms with FASD, resulting in misdiagnoses and/or missed diagnosis of FASD. This is amplified for Black youths due to the later onset of referral for assessment and lower frequency of referral to specialty clinics. This clinical case report depicts a misdiagnosis and a missed diagnosis of FASD in a 10-year-old African American patient, who was referred for neuropsychological evaluation. Diagnoses at the time of referral included attention-deficit/hyperactivity disorder (ADHD), oppositional defiant disorder (ODD), and disruptive mood dysregulation disorder (DMDD). Upon completion of a comprehensive evaluation, the patient's diagnoses changed to neurodevelopmental disorder associated with prenatal alcohol exposure, intellectual disability (ID), ADHD, and unspecified depressive disorder, leading to referral to appropriate interventions. The goal of this clinical case report is to increase clinician understanding of FASD and its clinical presentation, inform clinicians about the diagnostic and systemic factors that contribute to misdiagnosis and missed diagnosis of FASD, and to demonstrate the importance of an accurate diagnosis of FASD. By depicting the diagnostic challenges in an African American youth, the authors hope to bring awareness to the racial and ethnic disparities in the diagnosis of neurodevelopmental disabilities, specifically FASD in minority youth.

Keywords: Prenatal alcohol exposure, fetal alcohol spectrum disorders, fetal alcohol syndrome, alcohol-related neurodevelopmental disorder, misdiagnosis, missed diagnosis, Black youths

Fetal alcohol spectrum disorder (FASD) is an umbrella term used to describe the set of conditions that result from prenatal alcohol exposure (PAE).[1] FASD includes fetal alcohol syndrome (FAS), partial fetal alcohol syndrome (pFAS), alcohol-related neurodevelopmental disorder (ARND), and alcohol-related birth defects (ARBD).[2] These conditions can cause structural changes in the brain, physical abnormalities, cognitive impairment, neurodevelopmental delays, socioemotional and behavioral problems, medical complications, and secondary disabilities (e.g., school failure, delinquency, incarceration).[2] FASD is the most common preventable cause of developmental disabilities and birth defects in the United States.[3,4] FASD is a major social, economic, and public health concern.[2,4] In 2014, the estimates of the prevalence of FASD were 0.5 to two per 1,000 births, and 4.5 per 1,000 births per year for pFAS, ARND, and ARBD.[2] Currently, the estimated prevalence for the entire spectrum is 10 per 1,000 births.[5]

Current research indicates that there are differences in prevalence rate for FASD across racial/ethnic groups.[6] Notably, it is estimated that seven percent of children with FASD have never been diagnosed and seven percent of children with FASD have been misdiagnosed.[5] Attention-deficit/hyperactivity disorder (ADHD), intellectual disability (ID), oppositional defiant disorder (ODD), conduct disorder (CD), reactive attachment disorder (RAD), and communication disorders have many common symptoms with FASD in neurocognitive domains, as well as academic, adaptive, behavioral, emotional, and social functioning domains.[7,8] Thus, the similarities in the clinical presentations between FASD and the noted diagnoses result in can misdiagnoses and/or missed diagnosis of FASD.

Stoler and Holmes (1999) analyzed the medical records of women and infants for prenatal alcohol effects and found underrecognition of the effects of alcohol, which subsequently led to underdiagnosis.[9] More recently, Chasnoff, Wells, and King (2015) assessed the rate of misdiagnosis and missed diagnosis of FASD within foster and adoption populations.[7] Diagnoses at the time of the referral commonly included ADHD, ODD, RAD, and posttraumatic stress disorder (PTSD). Following comprehensive and multidisciplinary diagnostic evaluations, results indicated a misdiagnosis rate of 6.4 percent and a missed diagnosis rate of 80.1 percent for FASD. Participants eventually diagnosed with FASD required significant changes in their interventions and therapy services.

Overall, evidence suggests that the mental health needs of minority children are largely unmet.[10,11] There are racial and ethnic disparities in screening, diagnostic assessment, and access to formal mental health services. Specifically, African American males are misdiagnosed with externalizing disorders, and neurodevelopmental disorder

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diagnoses are missed. For example, Black youths are more likely to be diagnosed with CD and ODD than their white counterparts.[12,13,14] This is amplified in the juvenile justice and foster care systems in which youth of color are overrepresented and diagnosed with more historically stigmatizing mental health disorders, which lead to poorer outcomes, compared to white youths.[15,16] Furthermore, evidence indicates that minority youths, including Black youths, are referred to neurodevelopmental specialty clinics less frequently for evaluation and treatment, delaying the age of diagnosis and implementation of effective interventions.[17] ADHD is 40 percent less likely to be diagnosed in Black males, and compared to white males, they are 32 percent less likely to receive psychiatric treatment for ADHD.[12] In addition, significant delays have been reported in the diagnosis of autism for African American children (7.9 years) compared to white children (6.3 years).[11] Overall, due to the diagnostic process for FASD requiring an interdisciplinary evaluation and documented mental health disparities, identifying FASD in minority youths requires not only an understanding of FASD symptoms, but also consideration of the systemic issues.

Misdiagnosis and/or missed diagnosis of FASD may lead to secondary disabilities, which are conditions that arise due to the adverse impact of the primary symptoms of FASD. Examples of secondary disabilities are school failure, delinquency, incarceration, and psychiatric problems, such as depression and substance use. Furthermore, it may lead to incorrect behavioral and pharmacological treatments, caregiver stress, and disrupted placements within the foster care system.[7,8,18] These factors could lead to less stability and poorer prognosis for individuals with FASD.

The goal of this clinical case report is to increase clinician understanding of FASD and its clinical presentation, inform clinicians about the diagnostic and systemic factors that contribute to misdiagnosis and a missed diagnosis of FASD, and demonstrate the importance of an accurate diagnosis of FASD. By depicting the diagnostic challenges an African American youth experienced, the authors hope to bring awareness to the racial and ethnic disparities in the diagnosis of neurodevelopmental disabilities, specifically FASD, in minority youths.

CASE REPORT

"DeShawn" was a 10-year-old African American male who was referred for a neuropsychological evaluation to an outpatient training clinic by Child Protective Services (CPS) for diagnostic clarification (i.e., question of FASD), to evaluate his academic functioning, and to determine appropriate placement. Previous diagnoses included ADHD, ODD, and DMDD. Medications at the time of the referral included atomoxetine (40mg), clonidine (0.1mg), olanzapine (5mg), quetiapine (50mg), and melatonin (3mg).

DeShawn's background history obtained from his CPS caseworker indicated that his biological parents used alcohol and other illicit drugs, and DeShawn's mother's consumption of alcohol during her pregnancy with him was confirmed. Presenting concerns included behavioral and emotional dysregulation, as well as impairments in his adaptive functioning. DeShawn's behavioral outbursts were often triggered by "being told no" and consisted of screaming, cursing, and physical aggression towards peers and adults (e.g., kicking and hitting, throwing items).

DeShawn was removed from his birth mother's custody at age four due to his mother driving under the influence of alcohol. He was placed in his grandmother's custody and reportedly viewed his grandmother as his maternal figure. At age nine, DeShawn was placed in foster care as a result of witnessing domestic violence in his home. Three months after placement in foster care (age 10), DeShawn was psychiatrically hospitalized due to a violent incident that took place in his first foster home. Two days after he was released to a second foster home, he engaged in self-endangering behaviors and set his mattress on fire, which led to him being readmitted to the hospital for stabilization. Upon release from the hospital, DeShawn was admitted to a residential mental health treatment facility, and six months later, he was transferred to an alternate residential mental health treatment facility due to aggressive and disruptive behaviors.

DeShawn displayed difficulties with developing and maintaining relationships due to aggression towards peers. He attended fourth grade and reportedly earned passing grades, yet experienced difficulties with reading. He required frequent redirection, was easily distracted, and sometimes fell asleep during class.

DeShawn's height and weight seemed appropriate for his age, and he was of average build. There were no indications of abnormal gait or gross motor issues. DeShawn was observed by the evaluator to have some facial features characteristic of children with FASD, such as low nasal bridge and slanted eye shape.Rapport was easily established with DeShawn, as he engaged in reciprocal conversation and answered the examiner's questions. He made minimal eye contact, displayed flat affect, and slumped in his chair. His speech was hard to understand. In addition, DeShawn frequently left his chair, ran and jumped around the room, talked excessively, interrupted the examiner, and attempted to start tasks before the examiner finished providing instructions. He required frequent redirection and repetition of instructions. A history of auditory and/or visual hallucinations, paranoia, and delusions was denied.

The Stanford-Binet, Fifth Edition, was used to assess DeShawn's intellectual functioning, which was within the moderately impaired/delayed range. In addition, DeShawn demonstrated deficits in adaptive skills, including difficulty with controlling his impulses, making decisions, developing and maintaining friendships, using community resources, and implementing self-helps skills such as bathing, dressing, and food preparation. Deshawn had very low academic achievement skills and significant learning delays, which was in line with his low cognitive functioning.

Consistent with the neurocognitive presentation of individuals with FASD, DeShawn demonstrated significant executive dysfunction (i.e., distractibility and difficulty with controlling his impulses, set shifting, and planning and organizing) and impaired verbal and visual memory. Nevertheless, results demonstrated that DeShawn recognized previously learned verbal information when provided with cues. DeShawn's language comprehension skills were better developed than his overall cognitive functioning, which explained his ability to understand and converse with minimal difficulty. Behaviorally, DeShawn exhibited significant hyperactivity, impulsivity, difficulty staying focused, aggression, conduct issues, and depression, which were consistent with the presenting concerns.

DeShawn's test results indicated overall impairment in his neurocognitive functioning, which was influenced, in part, by prenatal

alcohol exposure, as alcohol consumption during pregnancy was confirmed. Thus, per the *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition*, (*DSM-5)* diagnostic criteria, he was diagnosed with neurodevelopmental disorder associated with prenatal alcohol exposure, which explained his low cognitive, academic, and adaptive functioning, as well as executive dysfunction.[19] DeShawn was also diagnosed with ID based on his low cognitive and adaptive functioning. In addition, his previous diagnosis of ADHD was confirmed, as ADHD and FASD are highly comorbid due to similar brain regions (e.g., frontal lobes) being impacted. DeShawn reported that he experienced low mood, loneliness, negative self-image, and difficulty with developing friendships. Although secondary disabilities, such as internalizing disorders, are often present in individuals with FASD, DeShawn's symptoms were not clinically significant; therefore, he was diagnosed with unspecified depressive disorder. DeShawn's previous diagnoses of ODD and DMDD were removed since the behavioral and emotional dysregulation he exhibited resulted from the adverse impact of prenatal alcohol exposure to brain regions presumed to mediate executive functioning.

DISCUSSION

Neurodevelopmental disorders often go unrecognized, leading to misdiagnoses and/or missed or delayed diagnosis, especially for minority youths.[7,20] As in this case, the patient was initially diagnosed with ODD and DMDD, and the confirmed prenatal alcohol exposure was not taken into consideration, thus missing the opportunity to evaluate for FASD. Subsequently, no referral to a specialty clinic was made, which is consistent with research indicating that Black youths are less likely to be referred to specialty clinics, subsequently delaying the initial evaluation and diagnosis of neurodevelopmental disorders.[21] Instead he received medication and therapy for ODD and DMDD to which he did not respond. He continued to exhibit behavioral outbursts and emotional dysregulation in the presence of consistent and clear directions and structure. Despite lack of progress, his behaviors continued to be perceived as noncompliance and defiance. This is often the experience of Black youths in which externalized behaviors are perceived as aggression and emotional dysregulation without consideration of contextual and environmental factors. However, in this case, once a referral was made for diagnostic clarification for FASD, the etiology of the disruptive behaviors and nuances of the presentation were considered. For example, DeShawn had difficulty learning from feedback and connecting behaviors to their consequences, which is common in individuals with FASD.[22] In addition, he had difficulty with staying focused and with set shifting, mental flexibility, goal-oriented thinking, learning rules, and perseverative thinking. Despite the presence of emotional dysregulation and noncompliance, results from the functional behavior assessment indicated that DeShawn did not experience persistent irritable mood between outbursts and that his noncompliance was a function of his neurocognitive deficits. Thus, the diagnoses of ODD and DMDD were removed as his emotional and behavioral dysregulation were better explained under the classification of FASD.

The patient's intellectual functioning was evaluated based on knowledge that cognitive functioning of individuals diagnosed with FASD is typically within the borderline range and that there is a need for a holistic understanding of presenting issues.[23,24] Furthermore, consideration of how cognitive functioning is evaluated in minority youths is central to appropriate clinical practice due to the historic misuse of cognitive testing results for this population.[25] In this case, DeShawn's cognitive and adaptive functioning were considered in light of the potential diagnosis of FASD due to the high comorbidity of ID and FASD.[26] The evaluator took care to identify an appropriate cognitive measure that suited the pateint's language, behavioral, and developmental needs and cultural context. Results from this comprehensive evaluation demonstrated moderately impaired intellectual functioning and deficits in adaptive skills, and thus a diagnosis of ID, moderate, was given. As in any other developmental disorder, intellectual functioning needs to be taken into consideration while choosing appropriate interventions to maximize treatment benefits. Thus, the missed diagnosis of ID could explain in part why prior interventions were not effective, since they were not tailored to his cognitive level.

The diagnoses of FASD and ADHD are highly comorbid, with up to 70 percent of youth diagnosed with FASD also meeting criteria for ADHD.[8,27] The clinical presentation of this comorbidity involves higher levels of impulsivity and greater difficulty with encoding information compared to youths with ADHD alone.[28] Despite being on ADHD medication during the evaluation, the patient demonstrated significant executive dysfunction, including distractibility and difficulty with simultaneous processing, mental flexibility, set shifting, controlling his impulses, and planning and organizing. Additionally, he exhibited significant hyperactivity, which required persistent redirection. Based on test results and behavioral observations, the previous diagnosis of ADHD was kept, as the patient's severity of executive dysfunction (i.e., cognitive and behavioral) was greater than typically exhibited in individuals diagnosed with FASD or ADHD alone.

FASD can also lead to secondary disabilities, which include academic underachievement and school failure, delinquency, and substance use.[29] Additionally, the emotional dysregulation associated with FASD increases the risk for internalizing disorders, such as depression, anxiety, and psychosis.[30,31,32] In this case, the patient experienced low mood, loneliness, and negative self-image, which warranted a diagnosis of unspecified depressive disorder. Consistent with the presentation of individuals with FASD, the patient had difficulty developing and maintaining friendships due to physical and verbal aggression, which contributed to his depressive symptoms. Overall, the addition of the unspecified depressive disorder diagnosis captured his presentation and allowed for further monitoring of and intervention for his secondary disabilities.

CONCLUSION

This case report is a representation of misdiagnosis and a missed diagnosis of a neurodevelopmental disorder, specifically FASD, for a Black youth. The patient presented to the clinic with diagnoses of ADHD, ODD, and DMDD at the age of 10 and a history of ineffective and inappropriate pharmacological and psychotherapy interventions. The patient received a comprehensive neuropsychological evaluation, which yielded the following diagnoses: neurodevelopmental disorder associated with prenatal alcohol exposure, ID, ADHD, and unspecified depressive disorder.

This case report highlights the importance of obtaining a comprehensive history that includes questioning the exposure to teratogens,

specifically alcohol, in utero and then considering the impact of the prenatal alcohol exposure on the developing brain. Adequate knowledge of the neuropsychological profile of FASD is imperative for clinicians as differential diagnoses are considered. In the case of FASD, many comorbid conditions mimic FASD, which can lead to misdiagnoses or missed diagnosis. This becomes more vital when working with Black youths since there is a tendency to attribute externalizing behaviors to disruptive behavior disorders despite contextual variables. Therefore, it is recommended that clinicians working in settings and populations with a higher incidence of FASD familiarize themselves with the clinical presentation to ensure accurate diagnosis and/or referral. It is also recommended that clinicians consider the potential for misdiagnoses when interventions are ineffective. Finally, it is recommended that clinicians working with Black youths diligently consider contextual factors when conceptualizing externalizing behaviors in order to refer youths to neurodevelopmental clinics for accurate diagnosis and treatment of FASD.

A major advantage of accurate and timely diagnosis (i.e., before the age of six) is the prevention of secondary disabilities.[29] Subsequently, children with FASD can receive appropriate and efficacious treatment in the form of medication adjustment, educational advocacy, and psychotherapy interventions to address attachment, behavioral, and sensory issues.[33,34] Another benefit of accurate diagnosis is the prevention of frequent displacement within the foster care system.[35] FASD is a major economic and public health concern and inaccurate diagnosis leads to inappropriate and ineffective interventions.[36] Thus, accurate diagnosis of FASD benefits the individual and the system.

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